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TYPES OF MUTATIONS AND THEIR POSSIBLE SIGNIFICANCE IN EVOLUTION.¹

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STATION FOR EXPERIMENTAL EVOLUTION

THE beginning of the twentieth century saw the rise of two concepts which have profoundly affected biological thought and been of increasing influence in the trend of experimental study of plants and animals. The mutation theory of deVries based on the evening primrose, and the laws of Mendel based on the garden pea, settled the date of birth of the modern science of genetics. The studies on these two plants have together formed a basis for the main bulk of our present genetic investigations. While the garden pea stands intimately associated with a conception of inheritance of wider application than was at first imagined, the evening primrose and the theory of mutation connected with it are by many considered to furnish an example of a valuable theory founded upon incorrect interpretations. The belief is growing that most of the new forms which have appeared in cultures of the *Oenotheras* are not mutations at all and that the evening primroses, as an abnormal group of plants, are not to be seriously considered as representative of the processes of evolution in normal forms.

In the short time at my disposal, I wish to outline some recent findings in the jimson weed (*Datura Stramonium*) which it is hoped may throw incidentally some light on the more highly involved phenomena in the *Oenotheras*, and which may serve as a basis of a brief discussion of their possible evolutionary significance.

The jimson weed is not supplied with a wide range of obvious Mendelian characters. The early studies of

¹ A paper presented before the American Society of Naturalists at the Chicago meeting, December 30, 1920.

Naudin (9) and Godron (7) as well as the later investigations of deVries (13), Bateson and Saunders (1) and the writer and Avery (4) on this species have shown that purple color in flower and stem is dominant to lack of purple in those parts, and that spiny capsules are dominant to smooth capsules. The writer with Avery (5) has been able to add a third pair of contrasting characters: "many nodes," causing tall stature, in contrast to "few nodes," causing low stature. These are all the allelomorphic pairs actually determined. Moreover, no variation has arisen in the writer's cultures during the last seven years' study of this species which gave evidence of differing from the present stock by a single Mendelian factor. Distinct variations, provisionally termed mutations, have, however, regularly recurred whenever a sufficiently large number of plants have been subjected to observation (5). So far as investigated, they have been found to be connected with a duplication of one or more of the normal chromosomes (6). The normal quota is 12 pairs: 12 being therefore the gametic, haploid, or x number; and 24 the somatic, diploid, or $2x$ number. The simplest type of duplication is the addition of an extra chromosome, probably by non-disjunction in one of the pairs of the diploid complement, giving $24 + 1$, or 25 chromosomes as the somatic number. In such plants there will be 11 sets of 2 homologous chromosomes each and 1 set of 3 homologous chromosomes.

We have on the chart, Table I, 12 recurrent mutants of the type just discussed; which, while perfectly distinct from each other and from the normal stock, have certain characteristics in common. All have been found to produce gametes with 12 *and* 13 chromosomes (therefore with 25 as the calculated somatic number); all have a relatively large proportion of bad pollen grains, varying in the different mutants from 8 per cent. in the Globe to 21 per cent. in the Spinach, as indicated in the chart; all fail to transmit the mutant complex to any considerable extent through the pollen, while they do

TABLE I

SOMATIC NUMBER OF CHROMOSOMES AND PERCENTAGE OF BAD POLLEN
FOUND IN NORMALS AND IN DIFFERENT MUTANTS

Types	Somatic Number of Chromosomes	Per Cent. Bad Pollen
NORMALS	24	2.7
MUTANTS		
Tetraploid ("New Species'")	48	3.3
Triploid	36	34.1
Simple Trisomic		
1. Globe	25	7.9
2. Poinsettia	25	12.9
2a. P. var. wiry	25	9.3
3. Cocklebur	25	18.3
4. Ilex	25	12.2
5. Mutilated	25	20.7
6. Sugar loaf	25	16.1
7. Rolled	25	8.4
8. Reduced	25	10.7
9. Buckling	25	10.4
10. Glossy	25	18.0
11. Microcarpic	25	12.8
12. Spinach	25	20.7

transmit it through the egg cells, although to only about one quarter of the offspring. That the offspring of these mutants repeat the parental type regularly in less than the 50 per cent. expected is probably due to the lessened vigor of growth of mutants in comparison with normals.

If the presence of an extra chromosome in a given set causes a specific mutation due to the constitution of this particular chromosome, rather than to the mere presence of an extra chromosome irrespective of its origin, there are at least two consequences to be expected. First there should be as many possible mutants of this type as there are chromosome sets which may undergo duplication. In other words there should be 12. Twelve, as a matter of fact, is the actual number which we had found before the nuclear condition had been determined. In addition, we have two or three mutant forms apparently belonging to this class for which it has not yet been possible to obtain chromosome counts. In appearance they are combinations or modifications of members of the

recurrent twelve. Secondly, it should be possible by breeding tests to connect up mutants with as many chromosome sets as there are known Mendelian factors, or factor groups. This connection we seem to have established between the mutant Poinsettia and the set of chromosomes which carries the factors for purple pigmentation in flower and stem.

TABLE II

TYPES OF CHROMOSOMAL DUPLICATION, GAMETIC AND SOMATIC FORMULAE FOR PLANTS HETEROZYGOUS FOR FACTOR PAIR A AND a AND RATIOS OBTAINED WHEN SUCH PLANTS ARE SELFED, TOGETHER WITH DIAGRAMS ILLUSTRATING THE CHROMOSOMAL CONDITION IN SOMATIC CELLS

No. of Extra Chromosomes in Set	No. of Sets Affected	Gametic Formula	Selfed Ratios	Somatic Formula	Somatic Diagram																										
2	12	AA + Aa AA + 4Aa + aa Aa + aa (12 + 12)	1A : 0a 35A : 1a 3A : 1a	AAAA AAaa Aaaa (12 + 12) + (12 + 12)																											
1	1	2A + a + AA + 2Aa A + 2a + 2Aa + aa 12, (12 + 1)	NOR. 8A : 1a MUT. 9A : 0a NOR. 5A : 4a MUT. 7A : 2a	} AAa } Aaa (12 + 12) + 1																											
1	12			(12 + 12) + 12																											
<div>No. of chromosomes 12 + Frequencies</div> <table><tr><td>0</td><td>1</td><td>2</td><td>3</td><td>4</td><td>5</td><td>6</td><td>7</td><td>8</td><td>9</td><td>10</td><td>11</td><td>12</td></tr><tr><td>1</td><td>12</td><td>66</td><td>220</td><td>495</td><td>792</td><td>924</td><td>792</td><td>495</td><td>220</td><td>66</td><td>12</td><td>1</td></tr></table>						0	1	2	3	4	5	6	7	8	9	10	11	12	1	12	66	220	495	792	924	792	495	220	66	12	1
0	1	2	3	4	5	6	7	8	9	10	11	12																			
1	12	66	220	495	792	924	792	495	220	66	12	1																			

The set of 3 chromosomes in the diagram, Table II, may be called the Poinsettia set, or the purple set. A Poinsettia plant may, to speak in terms of the dominant factor, be considered nulliplex with no dominant genes, or simplex, duplex or triplex with, respectively, 1, 2, or 3 dominant factors. There are therefore two types of heterozygotes, and under greenhouse conditions these apparently can be distinguished from each other as well as

from the homozygous dominants by different intensities of pigmentation. Simplex heterozygotes when selfed throw offspring with 5 dominants to 4 recessives among the normals, and 7 dominants to 2 recessives among the Poinsettias; while duplex heterozygotes should give a ratio of 8:1 among the normals, and all dominants among the Poinsettias. There is evidence which seems to indicate that the mutant Cocklebur is conditioned by duplication in the chromosome set which carries both the factors for spiny capsules and also those for number of nodes. If this is actually the case, we must assume that these two factor pairs are loosely linked in the same chromosome with about 50 per cent. crossing over, since they appear to segregate independently of each other.

We have been discussing duplication of a single member in only one of the 12 chromosome sets. On the lower part of the chart (Table II) is represented the only plant we have yet found with an extra chromosome in every one of its 12 sets. Such a plant is triploid. What its breeding behavior will be, can not be told before another season. If the chromosomes assort at random, the gametes theoretically should have the chromosome numbers indicated in the chart, and the counts which my colleague, Mr. Belling, has made from figures in pollen mother cells are not inconsistent with the distribution of the theoretical frequencies. One might expect such triploid plants to give rise to individuals intermediate between triploids and mutants of the Poinsettia type; in other words to mutants with duplication of chromosomes in 2, 3, 4, etc., up to duplication in all the 12 sets. Such compound mutants we have not yet been able to surely identify in our cultures; but we have never before this past season had a triploid plant, which from the wide range of gametic types in its egg cells would seem a likely source of such mutations.

Tetraploid plants have been discussed at yesterday's session of the Botanical Society of America. They represent a further duplication over those of the triploids

already mentioned in that there are 4 homologous chromosomes in each set in somatic cells. The homologous chromosomes therefore form tetrasomes, to use a new term,² instead of disomes as in normals or trisomes as in triploid plants. Members of these tetrasomes apparently assort at random in the reduction division. In consequence, certain peculiarities in breeding behavior result. Plants duplex for a dominant factor (AAaa) will, when selfed, give a ratio of 35 dominants to 1 recessive in the offspring. Plants simplex for the dominant (Aaaa) will give a 3:1 ratio in their offspring; but a third of the dominant offspring will throw 35:1 ratios in the next generation. Plants triplex for the dominant (AAAa) will give in the immediate offspring all dominants; one quarter of which, however, may be expected to give a 35:1 ratio in a later generation. The results expected from selfing the 5 zygotic types are shown in Table III.

It might be expected that mutant forms would be found in which doubling of the chromosomal number had involved only a single one of the 12 sets. Such mutants would bear the same relation to tetraploid plants with all the sets involved that the Poinsettia type of mutants bear to triploid plants. They have not yet been found, however.

² The following terms are suggested to designate sets with numbers of chromosomes from 1 to 12: monosome, disome, trisome, tetrasome, pentasome, hexasome, heptasome, oktasome, enneasome, dekasome, hendekasome, dodekasome.

The number of sets affected by duplication may be indicated by the terms: simple, double, triple, quadruple, quintuple, sextuple, septuple, octuple, nonuple, decuple, undecuple, duodecuple.

The Poinsettia and Globe are simple trisomic mutants. If the Globe and Poinsettia could be combined to form a mutant with 3 chromosomes each in two of the 12 sets, such a mutant would be called a double trisomic mutant. If differential viability of gametes does not interfere, the triploid plant already mentioned should produce, theoretically, offspring of all the trisomic types from simple to duodecuple. Haploid, diploid, triploid, tetraploid, etc., are terms already employed to designate plants with the same number of chromosomes in all the sets.

TABLE III

TETRAPLOID PLANTS. RESULTS OF SELFING TETRAPLOID PLANTS ARISING FROM THE CROSS OF A HOMOZYGOUS DOMINANT (AAAA) BY A RECESSIVE (aaaa), CARRIED TO THE F₄ GENERATION.

In the F₄, only phenotypes are represented.

P ₁ —	AAAA and aaaa							
F ₁ —AAaa	(Gametes of F ₁ — AA + 4Aa + aa)							
F ₂ —1AAAA	+	8AAaA	+	18AAaa	+	8Aaaa	+	1aaaa
F ₃ —AAAA	AAAA + 2AAaA + AAaa				AAaa + 2Aaaa + aaaa			aaaa
F ₄ —A	A	A	35A: 1a		35A: 1a	3A: 1a	a	a

It is possible that a single set in an otherwise tetraploid plant may have an extra chromosome, giving 5 chromosomes in one set and 4 in the remaining eleven. At least we have a single plant in a tetraploid pedigree which strongly resembles the Globe—the best known of our simple trisomic mutants. The cytological evidence shows that its chromosomal number is at least tetraploid, but is not yet sufficient to prove that its Globe-like appearance is determined by the addition of a fifth member to the chromosomal set responsible for the Globe mutant.

The occurrence of mutations of the types discussed in the foregoing paragraphs is bound up with the causes of chromosomal duplication. Knowing the mechanism to be affected, we may be able ultimately to induce chromosomal mutations by the application of appropriate stimuli.

We have outlined the types of chromosomal duplication already found in the jimson weed, and have shown some of the peculiarities in the breeding behavior of the mutant forms which they condition. It will be well to consider for a moment this process of duplication as it affects the individual plant and as it may have a possible significance in our theories of mutation and evolution.

The mutants of the Poinsettia or Globe type, in which but a single chromosomal set is involved in the duplication, should enable one to discover something in regard

to the influence of each specific chromosome upon the morphology and physiology of the datura plant. While there seems to be but a single chromosomal set responsible for the presence or absence of purple pigmentation, probably each chromosome has an influence upon the strength of expression of the pigment since the several mutants appear to differ widely in color when homozygous for the main purple factor. Thus Glossy is darker purple than normals, while Cocklebur is distinctly lighter. In normal plants there is a balanced adjustment between the modifying factors in the different chromosomes. When this balance is disturbed by the addition of only a single extra chromosome to one of the 12 sets, profound changes are brought about in the ontogeny of the resultant plant. When all of the sets have an extra chromosome, however, as is the case in triploids, no great disturbance of the balance is brought about and the plant is not greatly different from normals. Even in tetraploid plants where all the sets are equally affected, although the total number of chromosomes is doubled, the difference from normal is not so great as in mutants of the Globe and Poinsettia series. The leaves of tetraploid plants, when carrying the factor for many nodes, may be distinctly larger than those of normals. Few-noded tetraploids, however, are less easily distinguished. The best diagnostic character has been the globose shape of the capsule, and yet plants known to be tetraploid from cytological evidence have been found this past season with capsules perfectly normal in appearance.

What is the bearing of the phenomena of chromosomal duplication in datura upon the mutation theory? In the first place, the mutants of the Globe type apparently correspond to the *lata* type of mutants in the *Oenothera*s in which an odd somatic chromosome has been determined, although in these *Oenothera* mutants no breeding evidence has been available to show that the peculiarities of mutant *lata* are due to the presence of an extra chromosome in any specific chromosomal set. Our tetraploid

mutant "New Species" corresponds to *Oenothera gigas* and is brought about by a doubling of the chromosome number. The color ratios in our tetraploid daturas indicate that *Oenothera nanella* is a Mendelian segregate and suggest that other of the *Oenothera* mutants which give monohybrid ratios in crosses may be of the same nature. Our evidence in regard to *O. nanella* comes from the occurrence of this mutant in cultures of *O. gigas*. DeVries (14) reports that certain races of *gigas* when selfed regularly produce from 1 to 2 per cent. *nanella* mutants, while certain pedigrees give monohybrid ratios which, on account of the lesser vitality of the recessive *nanellas*, show a higher proportion of the dominant *gigas* forms. From the pedigrees approaching a 3:1 ratio he obtained plants which bred true, except again for the 1 to 2 per cent. of *nanella* mutants in their offspring. A glance at the chart (Table III) will show that, if our theory of tetraploidy be correct, the 1 to 2 per cent. of mutant *nanellas* which deVries obtained by selfing plants from 3:1 pedigrees must have been the recessives in a 35:1 ratio since no dominant plants in a 3:1 pedigree of a tetraploid race could be expected to breed true. The dominant phenotypes must either throw 3:1 ratios again or 35:1 ratios. The deviations of the *nanella* mutants in this case from a 35:1 ratio is accounted for by a similar proportionate deviation in the 3:1 ratio. The work of Muller (8) on balanced lethals strongly suggests that such of the *Oenothera* mutants as are not caused by chromosomal duplication are due to cross-overs from a balanced lethal condition.

What then is a mutation? I do not feel we need to be bound by its application to the evening primrose for reasons of priority, since Waagen (15) had previously used the term in paleontology in an entirely different sense. I believe, with the idea that mutations must involve a qualitative change, that we shall ultimately confine the term to mutations of genes, although such mutations may later be shown to be as different from our

present conceptions of them as are mutations in the *Oenotheras* from the conceptions in deVries's classical publication, "The Mutation Theory." It may still be desirable to employ the word *mutation* as a collective term to designate the sudden appearance of any apparent genetic novelty—whatever its real cause—until we know better. Strictly speaking I should not call chromosomal aberrations mutations when the changes are purely quantitative. The occurrence of tetraploidy would therefore be no more a mutation than the doubling of chromosomes at the origin of the sporophyte from the gametophyte of ferns.

We have seen that chromosomal duplications and related phenomena may simulate gene mutations in their effects upon the individual. What is their possible significance in evolution? Let us first consider tetraploidy. Numerous investigators have called attention to the fact that the chromosome numbers of plants are more frequently in multiples of two and four than one would expect from random sampling. Pairs of related species have been listed for which one member had twice as many chromosomes as the other. Such species have even been called tetraploid. We feel strongly the desirability of confining the term tetraploidy to those cases in which the $4x$ number is brought about by a doubling of homologous chromosomes. Doubling by transverse division is a possible method, but would not be included in the term.

Tetraploidy has been observed in experimental cultures of *œnothera*, *primula* and *datura*. Do such tetraploid plants occur in nature, and are they capable of giving rise to taxonomically new species? It may be mentioned that the tetraploid *datura* was called "New Species" before its tetraploid nature was suspected. It satisfied the requirements of an independent species. The pollen was relatively good, and the mutant formed a distinct race, self-fertile and fertile *inter se*, while practically sterile with the parent stock. Tetraploid plants, therefore, stand slight chance of being swamped by hybridization with the

species from which they have sprung. Once arisen, their chances of survival would depend upon their ability to complete with other forms in the struggle for existence. There are no certain cases of tetraploidy known outside of cultivation. It must be admitted, however, that their identification would be difficult. I have shown that gigantism is not an invariable diagnostic feature of tetraploid daturas. As yet no cytological criteria of tetraploidy have been established. The breeding behavior, which is the only safe test, might easily be misinterpreted, as it was apparently by deVries in the case of the tetraploid *gigas* and *nanella*. Moreover, a suspected form must show a pair of Mendelizing characters before a breeding test can be applied.

Despite the paucity of evidence in regard to the occurrence of tetraploidy in nature, the speaker believes that it may have been one of the principal methods in the evolution of plants. Its occurrence would furnish the barrier between a new species and its parental form that Darwin sought, and it would give a reason for the prevalence of even numbers in the counts of chromosome pairs. I believe that a search for tetraploid forms in nature will be rewarded. Perhaps they will more likely be found in horticultural races propagated by vegetative means. I take this occasion to suggest the desirability of testing for tetraploidy any *gigas*-like plant that may be found in the wild or under cultivation. We are making a special study of tetraploidy at the Station for Experimental Evolution and should be glad to receive plants suspected of being tetraploid from any who do not care to make the necessary tests themselves.

Even if proper tests should show that few forms in nature were tetraploid in the sense that each chromosomal set in somatic cells was composed of 4 homologous members, tetraploidy might still be a stage in the origin of species with an even number of pairs of chromosomes. In the 3 forms in which tetraploid plants have arisen under observation, the 4 homologous chromosomes in a

set apparently assort at random in the reduction division. If, instead of acting individually without predilection one for another, the four should come to assort in pairs, we should have a different ratio in the F_2 generation (15:1 instead of 35:1). There would still be duplication of genes and a 4x number in reference to the parental form, but independent assortment of the chromosomes would have been lost. It will not be possible to go into the details of the argument. It is at least suggestive that Shull (12) has found 3 cases of duplicate genes in the shepherd's purse, which has 32 chromosomes (that is 4 times 8); and Nilson-Ehle (10) has found a case of triplicate genes in a wheat having 42 chromosomes, which is 3 times the number in another variety of wheat (11).

If tetraploid plants have been of influence in evolution, it is probable that the other types of duplication have also been of influence. A mutant of the Globe type with a single duplication in one of the 12 sets ordinarily fails to hand on the duplication through the pollen. Occasionally it might do so, and we should then expect a constant race with 4 homologous chromosomes in one of the 12 sets. If these 4 should cease to assort at random and pair, we should again have a possible duplication of genes and an added pair of chromosomes characteristic of the race.

There is not time at my disposal to discuss mutations of genes. In a recent paper (2) on a somatic mutation in portulaca, I have indicated my belief that mutations of genes may occur at any stage in the development of the plant. We have found color mutations which affected only the epidermis, and therefore could not be transmitted through seeds. We have also found similar color mutations which affected only the sub-epidermal tissue, and therefore could not show in the petal; but which became evident from the seeds produced from this mutated tissue. There seems to be no preferred location for the origin of factorial mutations in flowering plants, although they are more readily transmitted if they occur in the

gametes or in the embryo. The fact that in vegetatively propagated *Mucors* (3) I have found mutations relatively common where the possibility of sexual reproduction was ruled out, indicates that sudden genetic changes are not necessarily associated with sexual processes.

It has not been possible in this brief presentation to give an extended classification of mutations, nor to discuss in detail their possible significance in evolution. It will be sufficient if I have made clear the distinction which must be kept in mind, in any discussion of the subject, between mutations in individual genes and those brought about by chromosomal aberrations.

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